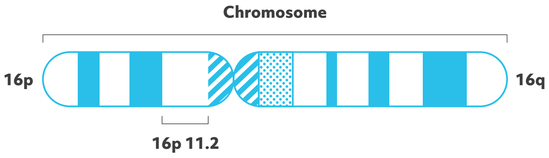
Chromosome 16p11.2 Copy Number Variants



- **Chromosomes** contain our genes. Most of us have 46 chromosomes, one set of 23 from mom and one set of 23 from dad.
- **Each chromosome has stripes, or bands**, that are unique to it and that can be seen using special laboratory techniques.
- **Chromosomes have arms.** Each chromosome has a short (or p) arm and a long (or q) arm.
- **Researchers have given genes addresses on the chromosomes.** The addresses indicate which chromosome the gene is on, what arm it is on, and what region of the chromosome it is in. 16p11.2 is on chromosome 16, arm p, region 11.2.

Sometimes a person is born with chromosomes that are different. One of the person's chromosomes may have extra genetic material or missing genetic material. When this happens, it is called a copy number variant, or CNV.

A 16p11.2 "duplication" means there is extra genetic material. A 16p11.2 "deletion" means that genetic material is missing. People with a duplication and people with a deletion are likely to have different features and symptoms.

16p11.2 has two distinct regions, the "proximal" and the "distal." The typical region is where duplications and deletions are most commonly found. The distal region is next to it.

You can find which section of your or your child's chromosome 16 is duplicated or deleted in your genetics lab report.

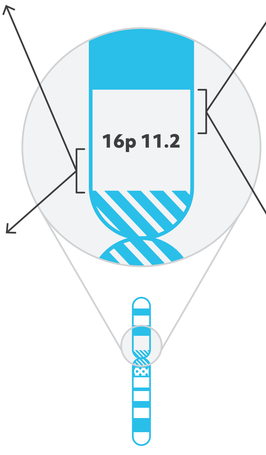
*Note: Mb is a genetic unit of length

BP4-BP5 DUPLICATION:
Extra genetic information appears in the ~29.5-30.1 Mb area.

A person with a BP4-BP5 duplication may have speech delay, developmental delay and intellectual disability, autism spectrum disorder, behavioral issues, and small head size.
Read more here.

BP4-BP5 DELETION:
Genetic information is missing from the area of ~29.5-30.1 Mb.

A person with a BP4-BP5 deletion may have speech and language disorders, developmental delay and intellectual disability, learning difficulties, autism spectrum disorder, behavioral issues, a tendency toward obesity, large head size, and seizures.
Read more here.



DISTAL DUPLICATION:
Extra genetic information appears in the ~28.8-29.1 Mb area.

A person with a distal duplication may have developmental delay and intellectual disability, autism spectrum disorder, speech delay, and seizures.

DISTAL DELETION:
Genetic information is missing from the area of ~28.8-29.1 Mb.

A person with a distal deletion may have developmental delay and intellectual disability, speech delay, autism spectrum disorder, behavioral issues, early feeding difficulties, and a tendency toward obesity.

Everyone is unique.

People with a 16p11.2 CNV exhibit different features. Even members of the same family may show a wide range of features.

We are still learning about 16p11.2.

With the help of families, we can gain a better understanding through research. Scientists have discovered a lot about 16p 11.2 with the help of families. That being said, we still do not know everything. If you would like to learn more, join our community, or contribute to the ongoing research, please visit **simonssearchlight.org**